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DEPARTMENT OF COMMERCE

National Institute of Standards and Technology

Genome in a Bottle Consortium – Progress and Planning Workshop

AGENCY: National Institute of Standards and Technology, Commerce.

ACTION: Notice of public workshop.

SUMMARY: The National Institute of Standards and Technology (NIST) announces the Genome in a Bottle Consortium - Progress and Planning Workshop to be held on Thursday, January 28, 2016, and Friday, January 29, 2016. The Genome in a Bottle Consortium is developing the reference materials, reference methods, and reference data needed to assess confidence in human whole genome variant calls. A principal motivation for this consortium is to enable performance assessment of sequencing and science-based regulatory oversight of clinical sequencing. The purpose of this workshop is to update participants about progress of the consortium work, continue to get broad input from individual stakeholders to update or refine the consortium work plan, continue to broadly solicit consortium membership from interested stakeholders, and invite members to participate in work plan implementation. Topics of discussion at this workshop will include progress and planning of the Analysis Group, which is analyzing and integrating the large variety of sequencing data for four

candidate NIST Reference Materials (RMs), with a particular focus on challenging types of variants and challenging regions of the genome. Other potential NIST RMs that might be developed by the consortium will also be discussed.

DATES: The Genome in a Bottle Consortium workshop will be held on Thursday, January 28, 2016 from 9:00 a.m. to 5:30 p.m. Pacific Time, and Friday, January 29, 2016 from 9:00 a.m. to 1:00 p.m. Pacific Time. Attendees must register by 5:00 p.m. Pacific Time on Thursday, January 21, 2016.

ADDRESSES: The meeting will be held on the second floor of the Li Ka Shing Conference Center, Stanford University, 291 Campus Drive, Palo Alto, CA 94305. Please note admittance instructions under the SUPPLEMENTARY INFORMATION section of this notice.

FOR FURTHER INFORMATION CONTACT: For further information contact Justin Zook by email at jzook@nist.gov or by phone at (301) 975-4133 or Marc Salit by email at salit@nist.gov or by phone at (650) 350-2338. To register, go to: http://web.stanford.edu/~saracl/GIAB2016.fb

SUPPLEMENTARY INFORMATION: Clinical application of ultra high throughput sequencing (UHTS) for hereditary genetic diseases and oncology is rapidly growing. At present, there are no widely accepted genomic standards or quantitative performance metrics for confidence in variant calling. These standards and quantitative performance metrics are

needed to achieve the confidence in measurement results expected for sound, reproducible research and regulated applications in the clinic. On April 13, 2012, NIST convened the workshop "Genome in a Bottle" to initiate a consortium to develop the reference materials, reference methods, and reference data needed to assess confidence in human whole genome variant calls (www.genomeinabottle.org). On August 16-17, 2012, NIST hosted the first large public meeting of the Genome in a Bottle Consortium, with about 100 participants from government, academic institutions, and industry. This meeting was announced in the Federal Register (77 FR 43237) on July 24, 2012. A principal motivation for this consortium was to enable science-based regulatory oversight of clinical sequencing.

At the August 2012 meeting, the consortium established work plans for four technical working groups with the following responsibilities:

(1) Reference Material (RM) Selection and Design: select appropriate sources for whole genome RMs and identify or design synthetic DNA constructs that could be spiked-in to samples for measurement assurance.

(2) Measurements for Reference Material Characterization: design and carry out experiments to characterize the RMs using multiple sequencing methods, other methods, and validation of selected variants using orthogonal technologies.

(3) Bioinformatics, Data Integration, and Data Representation: develop methods to analyze and integrate the data for each RM, as well as select appropriate formats to represent the data.

(4) Performance Metrics and Figures of Merit: develop useful performance metrics and figures of merit that can be obtained through measurement of the RMs.

The products of these technical working groups will be a set of well-characterized whole genome and synthetic DNA RMs along with the methods (documentary standards) and reference data necessary for use of the RMs. These products will be designed to help enable translation of whole genome sequencing to regulated clinical applications. The pilot, NIST "Human DNA for Whole-Genome Variant Assessment (Daughter of Utah/European Ancestry)" RM was released in May 2015 and is available at http://tinyurl.com/giabpilot. The consortium is currently analyzing and integrating data from two trios that are candidate NIST RMs. The consortium meets in workshops two times per year, in January at Stanford University in Palo Alto, CA, and in August at the National Institute of Standards and Technology in Gaithersburg, MD. At these workshops, including the last meetings at Stanford in January 2015 and at NIST in August 2015, participants in the consortium have discussed progress developing well-characterized genomes for NIST Reference Materials and planned future experiments and analysis of these genomes (see https://federalregister.gov/a/2012-18064, https://federalregister.gov/a/2013-18934, https://federalregister.gov/a/2014-18841 and https://federalregister.gov/a/2015-01158 for past workshops at NIST and Stanford). The January 2015 meeting was announced in the Federal Register (80 FR 3220) on January 22, 2015, and the meeting is summarized at https://docs.google.com/document/d/19J6YDg1MH1iD-8Q8mmV9L7wHOfuyUC3aogctZ2Nh87U/edit?usp=sharing. The August 2015 meeting was announced in the Federal Register (80 FR 45194) on July 29, 2015, and the meeting is summarized at https://docs.google.com/document/d/19-KSn0ydF8rsWRbl6OqhIdbt2gGN10dOMRF6inKmrk4/edit?usp=sharing.

There is no cost for participating in the consortium. No proprietary information will be shared as part of the consortium, and all research results will be in the public domain.

All attendees are required to pre-register. Anyone wishing to attend this meeting must pre-register at http://web.stanford.edu/~saracl/GIAB2016.fb by 5:00 p.m. Pacific Time on Thursday, January 21, 2016, in order to attend.

Dated:

Richard Cavanagh
Acting Associate Director of Laboratory Programs
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